About haemophilia

Haemophilia is caused by either a deficiency or a complete lack of one of the blood clotting factors. As a result, the blood does not clot completely, causing patients with haemophilia to experience bleeding, especially in the joints. Frequent bleeding may result in joint damage and physical disabilities, significantly limiting the patient's quality of life.

There are different types of haemophilia. The most common form, haemophilia A, is caused by a clotting factor VIII (FVIII) deficiency. The rarest form of the disorder, haemophilia B, is caused by a clotting factor IX deficiency. The effects of both forms are the same, however.

Haemophilia is a hereditary disease that is transmitted through the X chromosome and therefore primarily occurs in males. Females with a defective FVIII or IX gene do not exhibit the bleeding typical in affected males because females have a second X chromosome and therefore a second FVIII or IX gene. Females with a defective FVIII or IX gene can pass the disease on to their children and are therefore called carriers of the disease.

If a mother is a carrier and the father is unaffected, there is a 50% chance that any son produced by the couple will be affected by haemophilia and a 50% chance that any daughter will be a carrier of the disease.[1]

Percentage of active factor VIII or IX in the blood[2]

50–150%	5–40%	1–5%	Less than 1%
Normal	Mild haemophilia	Moderate haemophilia	Severe haemophilia

Frequency of haemophilia

Haemophilia occurs in one in 10,000 births.[2] Severe haemophilia is usually diagnosed within the first year of life.

Haemophilia worldwide[2]

Approximate number of people with diagnosed haemophilia in the world:

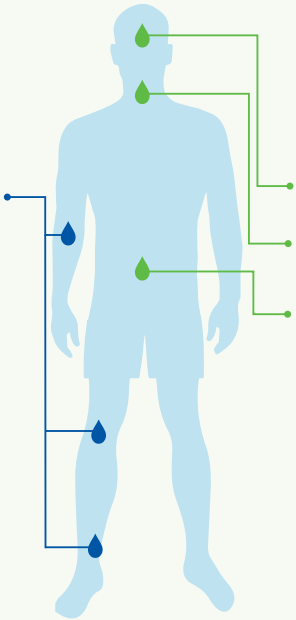
400,000

Where most bleeds occur[2]

Severe bleeds

In muscle
10–20%

In the joints (ankles, knees and elbows)
70–80%



Life-threatening bleeds

Intracranial

Throat/mouth

Gastrointestinal
5–10%

A **target joint** is a joint that is affected by three or more spontaneous bleeds within a period of six consecutive months.

Treatment options

Haemophilia is treated with 'replacement therapy'. The missing clotting factor is introduced into the blood. Factor replacement is either taken from human plasma or produced using recombinant technologies.[3]

Prophylaxis
This therapy involves regularly injecting factor VIII or IX in order to prevent bleeding.

On-demand care
This therapy involves treating only acute bleeds in an effort to stop bleeding as quickly as possible.

Early and regular prophylaxis with recombinant factor VIII in children younger than six years old with haemophilia A has shown a reduced risk of joint damage by 84% compared to on-demand care.[4]

Surgery/clinic stamp

Emergency telephone

Further useful information on haemophilia can be found on the World Federation of Hemophilia (WFH) website.

http://www.wfh.org

1 *"What Causes Hemophilia?" NIH, National Heart, Lung, and Blood Institute. July 2013. Accessed October 24, 2014 from http://www.nhlbi.nih.gov/health/health-topics/topics/hemophilia/causes.html.*

2 *Srivastava, Dr. Alok, et al. Guidelines for the Management of Hemophilia. 2nd ed. Montreal: Blackwell, 2012.*

3 *Lee, Christine A., Erik E. Berntorp and W. Keith Hoots, eds. Textbook of Hemophilia. 2nd ed. Blackwell, 2010.*

4 *Manco-Johnson, Marilyn J., et al. "Prophylaxis versus Episodic Treatment to Prevent Joint Disease in Boys with Severe Hemophilia." The New England Journal of Medicine (2007). Accessed October 24, 2014 from http://www.nejm.org/doi/full/10.1056/NEJMoa067659.*

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